



PRKRA gene

protein activator of interferon induced protein kinase EIF2AK2

Normal Function

The *PRKRA* gene provides instructions for making a protein called PACT. This protein plays a role in the cell's response to stress, such as exposure to viruses, damaging molecules called free radicals, or other toxic substances. When a cell is under stress, the PACT protein turns on (activates) another protein called PKR, which then turns off (inactivates) the eIF2 alpha (eIF2 α) protein. Inactivation of eIF2 α lowers protein production, which helps protect cells from damage. The signals triggered by PACT can ultimately lead to self-destruction (apoptosis) of the cell if it remains under stress.

The signals sent by the PACT protein are also important for a process in the brain called synaptic plasticity. Synaptic plasticity is the ability of the connections between brain cells (synapses) to change and adapt over time in response to experience. This process is critical for learning and memory.

Health Conditions Related to Genetic Changes

Dystonia 16

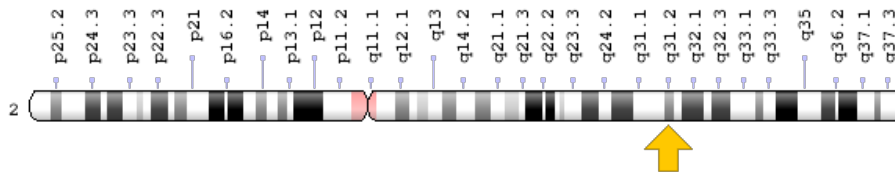
At least eight mutations in the *PRKRA* gene have been identified in people with dystonia 16. This condition is one of many forms of dystonia, which is a group of conditions characterized by involuntary movements, twisting (torsion) and tensing of various muscles, and unusual positioning of affected body parts.

Most of the *PRKRA* gene mutations that cause dystonia 16 change single protein building blocks (amino acids) in the PACT protein. Studies suggest that these changes alter the timing of signals sent by the PACT protein in response to stress. The abnormal signaling increases the rate at which cell death occurs. It is likely that the excessive loss of cells in certain regions of the brain impairs the brain's ability to control muscles and movement, resulting in the features of dystonia 16. It is unclear why brain cells are particularly affected by *PRKRA* gene mutations.

Chromosomal Location

Cytogenetic Location: 2q31.2, which is the long (q) arm of chromosome 2 at position 31.2

Molecular Location: base pairs 178,431,414 to 178,451,175 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DYT16
- PACT
- PRKRA gene
- protein activator of the interferon-induced protein kinase

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The Phosphorylation of an Initiation Factor Globally Regulates Protein Synthesis
https://www.ncbi.nlm.nih.gov/books/NBK26890/#_A1387_

Clinical Information from GeneReviews

- Hereditary Dystonia Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1155>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28PRKRA%5BTIAB%5D%29+OR+%28DYT16%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- PROTEIN KINASE, INTERFERON-INDUCIBLE DOUBLE-STRANDED RNA-DEPENDENT ACTIVATOR
<http://omim.org/entry/603424>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PRKRA.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PRKRA%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:9438
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:8575>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8575>
- UniProt
<https://www.uniprot.org/uniprot/O75569>

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<https://ghr.nlm.nih.gov/gene/PRKRA>

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